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SENATE

{ REPORT
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OFFICE FOR RARE DISEASE RESEARCH ACT OF 1995

MAY 4 (legislative day, MAY 1), 1995.—Ordered to be printed

Mrs. KASSEBAUM, from the Committee on Labor and Human Resources, submitted the following

REPORT

[To accompany S. 184]

The Committee on Labor and Human Resources to which was referred the bill (S. 184) to establish an Office for Rare Disease Research in the National Institutes of Health, and for other purposes, having considered the same, reports favorably thereon without amendment and recommends that the bill do pass.

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I. SUMMARY OF THE BILL

The legislation codifies the Office for Rare Disease Research within the Office of the Director of the National Institutes of Health (NIH). The Director of the National Institutes of Health will appoint a Director of the Office.

The purpose of the Office is to promote and coordinate research on rare disease through the development of a strategic research plan and a clinical research database. The act establishes an advisory council to provide advice for carrying out the activities of the Office to the Director of the Office for Rare Disease Research.

The Director of the Office will develop a strategic research plan; disseminate and coordinate information among the Institutes and the public; support research training; identify research opportunities for the NIH; develop and maintain a research database on current government sponsored clinical research; determine the need for registries of research subjects and epidemiological studies of rare disease populations; and prepare biennial reports on the activities of the Office and submit them to the Secretary and the Congress.

II. BACKGROUND AND NEED FOR LEGISLATION

There are over 5,000 rare diseases. Between 10 and 20 million Americans suffer from such diseases in this country. Each rare disease affects less than 200,000 Americans. These illnesses can be as devastating to those afflicted as cancer or heart disease are to larger numbers of patients. Marfan's syndrome, Supranuclear palsy, Cystic Fibrosis, Addison Disease, Duchenne's Muscular Dystrophy, and Sarcoidosis are a few examples of rare diseases that have a profound impact on the quality of life and life expectancy.

Most physicians are unfamiliar with these diseases, and early diagnosis is often missed. Sometimes patients must travel across the country for treatment, since few physicians are trained in treating such diseases.

The National Institutes of Health's history of sponsoring rare disease research is well established and recognized. Each institute supports research on rare diseases. For example, the National Institute of Allergy and Infectious Diseases supports research on numerous diseases that are classified as rare—including Hemophilus influenzae, Lyme Disease, Chronic Fatigue Syndrome, and Hepatitis C. The National Institute of Child Health and Human Development supports research in the prevention, diagnosis, evaluation and treatment of many rare diseases and disorders including PKU, cystic fibrosis, sudden infant death syndrome, osteogenesis imperfecta, cystinosis and Alpha 1-Antitrypsin Deficiency. The National Heart, Lung and Blood Institute supports research on rare diseases such as cardiomyopathies, cystic fibrosis, sudden and unexplained death in sleep, familial lipid disorders, Raynaud's Syndrome, pulmonary hypertension, sickle cell anemia, Cooley's anemia and Fanconi anemia. In addition, the NIH supports research in Wilson's Disease, Wilm's Tumor, Osteosarcoma, Hairy Cell Leukemia, Juvenile Rheumatoid Arthritis, Fragile X Syndrome, Sjogren's Syndrome, Amyotrophic Lateral Sclerosis, Multiple Chemical Sensitivity and many other rare diseases.

A major concern of the rare disease community is the lack of easy access to the latest research findings and the location of ongoing and planned rare disease research studies. There are many institutes at the NIH, each charged with undertaking research in its respective field, with little coordination among them on rare disease. Many rare diseases affect multiple organ systems, so research on those diseases may be in the jurisdiction of several institutes. For example, scleroderma is a connective tissue disease of unknown origin, characterized by excessive collagen deposition in the skin and various vital organs including lung, heart and GI tract. The skin manifestations are the responsibility of the National Institute

of Arthritis and Musculoskeletal and Skin Diseases; the pulmonary and cardiac manifestations fall under the National Heart, Lung and Blood Institute; the gastrointestinal complications are investigated by the National Institute of Diabetes and Digestive and Kidney Diseases; and genetic research may be pursued at the Human Genome Center.

The question before Congress is one of how to coordinate government-sponsored research on rare diseases in such a way as to maximize efficiency by reducing unnecessary duplication. The Office for Rare Disease Research at the NIH will coordinate the activities of all the Institutes and execute programs with a minimal waste of time and funds.

The purpose of this legislation is to provide a focal point within the NIH to draw attention to rare diseases and to fashion a consensus for a strategic research vision and a clinical database which will enhance ongoing research in rare diseases. The establishment of a central Office will result in an inventory of the rare disease research currently being conducted at the NIH and will uncover opportunities for further development as they occur.

III. LEGISLATIVE HISTORY AND COMMITTEE ACTION

S. 184 was introduced on January 9, 1995, by Senator Hatfield. The bill was referred to the Committee on Labor and Human Resources.

In the executive session of the Committee on Labor and Human Resources held on Wednesday, March 29, 1995, S. 184 was brought up for consideration. The bill was adopted by voice vote and favorably reported to the full Senate.

IV. COMMITTEE VIEWS

The committee recognizes that between 10 and 20 million Americans suffer from rare diseases. The plight of those who endure these rare diseases is compounded by the reality that, oftentimes, little or no research is being conducted into the causes, treatments, and cures of these diseases. Those with rare diseases must maneuver among what seems an endless morass of fragmented federal agencies in the hope of receiving information about ongoing research on their specific ailment, as rare disease research is frequently underrepresented in the private sector.

The Office is charged with formulating a strategic research plan by which to execute rare disease research. This comprehensive plan is designed to bring about cooperation among the 20 institutes of the NIH. The Office is directed to identify current research supported by the federal government, opportunities and the needs and priorities for rare disease research, so as to provide each Institute with sufficient relevant information about ongoing research to prevent unintended duplication. In addition, the Office can make recommendations about where projects may be coordinated and/or consolidated. These activities will be overseen by the Director of the Office in consultation with the Advisory Council established in the legislation.

The legislation also calls for a database of ongoing government-sponsored clinical research projects. This database, which the com-

mittee encourages be made accessible to the public, will make it possible to connect researchers with patients for clinical trials, provide physicians and individuals with information on trials, and connect patients with support groups—a vital part of an individual's physical and psychological coping with rare disease.

The committee believes that the greatest hope for people suffering with rare diseases is the possibility of medical breakthroughs achieved through biomedical research.

V. COST ESTIMATE

U.S. CONGRESS,
CONGRESSIONAL BUDGET OFFICE,
Washington, DC, April 26, 1995.

Hon. NANCY LANDON KASSEBAUM,
*Chairman, Committee on Labor and Human Resources,
U.S. Senate, Washington, DC.*

DEAR MADAM CHAIRMAN: The Congressional Budget Office has prepared the enclosed cost estimate for S. 184, the Office for Rare Disease Research Act of 1995, as ordered reported by the Senate Committee on Labor and Human Resources on March 29, 1995.

Enactment of S. 184 would not affect direct spending or receipts. Therefore, pay-as-you-go procedures would not apply to this bill.

If you wish further details on this estimate, we will be pleased to provide them.

Sincerely,

JAMES L. BLUM
(For June E. O'Neill, Director).

Enclosure.

CONGRESSIONAL BUDGET OFFICE COST ESTIMATE

1. Bill number: S. 184.
2. Bill title: Office for Rare Disease Research Act of 1995.
3. Bill status: As ordered reported by the Senate Committee on Labor and Human Resources on March 29, 1995.
4. Bill purpose: S. 184 would establish an Office for Rare Disease Research within the National Institutes of Health (NIH).
5. Estimated cost to the Federal Government: The following table summarizes the estimated costs of establishing and operating an Office for Rare Disease Research, assuming appropriations of the necessary amounts.

FEDERAL GOVERNMENT COSTS

[By fiscal year, in millions of dollars]

	1996	1997	1998	1999	2000
Estimated authorizations of appropriations	1	1	2	2	2
Estimated outlays	(¹)	1	1	2	2

¹ Less than \$500,000.

The costs of this bill fall within budget function 550.

6. Basis of estimate: An office currently exists that meets some of the requirements in the bill and is funded at \$150,000 for fiscal year 1995. The bill would require the office to support training in the conduct of research on rare diseases, which is not done by the

existing office. According to NIH, the average cost of a three-year research fellowship is \$60,000 per year. If the Office for Rare Disease Research funded 5 research fellowships per year, the cost would be \$300,000 in fiscal year 1996, increasing to \$1 million per year for fiscal years 1998 through 2000. In addition, the bill would require the office to establish and maintain a clinical research database. CBO estimates that this effort would cost about \$600,000 in 1996 and about \$500,000 in subsequent years. Disseminating information and establishing an advisory council would cost \$150,000 annually.

This estimate assumes that all authorizations are fully appropriated at the beginning of each fiscal year. Outlays are estimated using spendout rates computed by CBO on the basis of recent program data for NIH.

7. Pay-as-you-go considerations: None.

8. Estimated cost to State and local governments: None.

9. Estimate comparison: None.

10. Previous CBO estimate: None.

11. Estimate prepared by: Murray Ross.

12. Estimate approved by: Robert A. Sunshine, for Paul Van de Water, Assistant Director for Budget Analysis.

VI. REGULATORY IMPACT STATEMENT

The committee has determined that there will be no increase in the regulatory burden or paperwork as the result of this bill.

VII. SECTION-BY-SECTION ANALYSIS

SECTION 1. SHORT TITLE

Section 1 provides that the act be cited as "Office for Rare Disease Research Act of 1955".

SECTION 2. ESTABLISHMENT OF OFFICE FOR RARE DISEASE RESEARCH

Section 2 amends part A of title IV of the Public Health Service Act (42 U.S.C. 281 et seq.) by adding a new Sec. 404F., Office for Rare Disease Research.

Subsection (a) establishes within the Office of the Director of the National Institutes of Health an Office for Rare Disease Research. The Director of the Office would be appointed by the Director of the National Institutes of Health.

Subsection (b) provides the purpose of the Office. The purpose of the Office is to promote and coordinate the conduct of research on rare disease through a strategic research plan and to establish and manage a rare disease research clinical database.

Subsection (c) requires the Secretary of Health and Human Services to establish an Advisory Council to provide advice to the Director of the Office for Rare Disease Research for carrying out the duties of the Office.

Subsection (d) requires the Director of the Office to develop a strategic research plan; disseminate and coordinate information among the Institutes and the public; support research training; identify research opportunities for the NIH; develop and maintain a research database on current government sponsored clinical re-

search; determine the need for registries of research subjects and epidemiological studies of rare disease populations; and prepare biennial reports on the activities of the Office and submit them to the Secretary and the Congress.

VIII. CHANGES IN EXISTING LAW

In compliance with rule XXVI paragraph 12 of the Standing Rules of the Senate, the following provides a print of the statute or the part or section thereof to be amended or replaced (existing law proposed to be omitted is enclosed in black brackets, new matter is printed in italic, existing law in which no change is proposed is shown in roman):

PUBLIC HEALTH SERVICE ACT

* * * * *

TITLE IV—NATIONAL RESEARCH INSTITUTES

PART A—NATIONAL INSTITUTES OF HEALTH

ORGANIZATION OF THE NATIONAL INSTITUTES OF HEALTH

SEC. 401. (a). * * *

* * * * *

SEC. 404F. OFFICE FOR RARE DISEASE RESEARCH.

(a) *ESTABLISHMENT.*—*There is established within the Office of the Director of the National Institutes of Health an office to be known as the Office for Rare Disease Research (in this section referred to as the "Office"). The Office shall be headed by a director, who shall be appointed by the Director of the National Institutes of Health.*

(b) *PURPOSE.*—*The purposes of the Office is to promote and coordinate the conduct of research on rare diseases through a strategic research plan and to establish and manage a rare disease research clinical database.*

(c) *ADVISORY COUNCIL.*—*The Secretary shall establish an advisory council for the purpose of providing advice to the director of the Office concerning carrying out the strategic research plan and other duties under this section. Section 222 shall apply to such council to the same extent and in the same manner as such section applies to committees or councils established under such section.*

(d) *DUTIES.*—*In carrying out subsection (b), the director of the Office shall—*

(1) develop a comprehensive plan for the conduct and support of research on rare diseases;

(2) coordinate and disseminate information among the institutes and the public on rare diseases;

(3) support research training and encourage the participation of a diversity of individuals in the conduct of rare disease research;

(4) identify projects or research on rare diseases that should be conducted or supported by the National Institutes of Health;

(5) develop and maintain a central database on current government sponsored clinical research projects for rare diseases;

(6) determine the need for registries of research subjects and epidemiological studies of rare disease populations; and

(7) prepare biennial reports on the activities carried out or to be carried out by the Office and submit such reports to the Secretary and the Congress.

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